

CLAIMS

1. An isolated DNA or gene:

comprising a full-length base sequence according to the sequence ID. No. 1 or 2, or a partial sequence thereof, or a base sequence hybridizable thereto or hybridizable with a complementary strand thereof, and being associated with Parkinson's disease.

2. An isolated DNA or gene:

comprising the base sequence of claim 1, or the full-length base sequence thereof, or the base sequence partially thereof, and the isolated DNA or gene whose gene defect is responsible for Parkinson's disease, or comprising a base sequence hybridizable thereto or hybridizable with a complementary strand thereof.

3. An isolated DNA or gene comprising the base sequence of claim 1 or 2, the isolated DNA or gene being variant thereof by alternative splicing, and being associated with Parkinson's disease, or the isolated DNA or gene comprising a base sequence hybridizable thereto or hybridizable with a complementary strand thereof.

4. A gene comprising the base sequence of any one of claims 1 to 3 whose gene product encodes a protein having a

substantially equivalent function to a protein comprising 1 to 465 amino acid sequence in the sequence ID. No. 1 or to a protein comprising 1 to 437 amino acid sequence in the sequence ID. No. 2.

5. An isolated DNA or gene comprising a gene which has caused an exonic deletion, a nonsense base substitute, a missense base substitute, a base deletion, a base addition, a base insertion, a splicing abnormality and/or a frameshift with respect to the base sequence of any one of claims 1 to 4; or comprising a base sequence hybridizable thereto or hybridizable with a complementary strand thereof, and the isolated DNA or gene being associated with Parkinson's disease.

6. An isolated DNA or a gene, or a gene fragment comprising a partial base sequence of the DNA or the gene of any one of claims 1 to 5, or an isolated DNA or a gene or a gene fragment comprising a base sequence hybridizable thereto or hybridizable with a complementary strand thereof.

7. A gene encoding a protein (a) or (b) comprising:

(a) the protein comprising 1 to 465 amino acid sequence in the sequence ID. No. 1;

(b) the protein in which one or more amino acid(s) of the amino acid sequence is or are deleted, substituted, or

added, and the protein being associated with Parkinson's disease.

8. A gene encoding a protein (c) or (d):

(c) the protein comprising 1 to 437 amino acid sequence in the sequence ID. No. 2;

(d) the protein in which one or more amino acid(s) of the amino acid sequence is or are deleted, substituted, or added, and the protein being associated with Parkinson's disease.

9. A recombinant vector comprising the DNA fragment or the gene of any one of claims 1 to 8.

10. A gene of claim 1:

comprising the full-length base sequence in the sequence ID. No. 1 such that eleven introns are intervened among twelve exons on the genome; and

encoding a protein having 1 to 465 amino acid sequence in a part (102 to 1496) of the base sequence.

11. A gene of claim 10 comprising the following base sequence of the intron in a boundary region between the exon and the intron:

the intron intervening between exon 1 and exon 2 has a

base sequence shown in the sequence ID. No. 3 adjacent to the 3' end of the exon 1, and has a base sequence shown in the sequence ID. No. 4 adjacent to the 5' end of the exon 2;

the intron intervening between exon 2 and exon 3 has a base sequence shown in the sequence ID. No. 5 adjacent to the 3' end of the exon 2, and has a base sequence shown in the sequence ID. No. 6 adjacent to the 5' end of the exon 3;

the intron intervening between exon 3 and exon 4 has a base sequence shown in the sequence ID. No. 7 adjacent to the 3' end of the exon 3, and has a base sequence shown in the sequence ID. No. 8 adjacent to the 5' end of the exon 4;

the intron intervening between exon 4 and exon 5 has a base sequence shown in the sequence ID. No. 9 adjacent to the 3' end of the exon 4, and has a base sequence shown in the sequence ID. No. 10 adjacent to the 5' end of the exon 5;

the intron intervening between exon 5 and exon 6 has a base sequence shown in the sequence ID. No. 11 adjacent to the 3' end of the exon 5, and has a base sequence shown in the sequence ID. No. 12 adjacent to the 5' end of the exon 6;

the intron intervening between exon 6 and exon 7 has a base sequence shown in the sequence ID. No. 13 adjacent to the 3' end of the exon 6, and has a base sequence shown in the sequence ID. No. 14 adjacent to the 5' end of the exon 7;

the intron intervening between exon 7 and exon 8 has a base sequence shown in the sequence ID. No. 15 adjacent to the

3' end of the exon 7, and has a base sequence shown in the sequence ID. No. 16 adjacent to the 5' end of the exon 8;

the intron intervening between exon 8 and exon 9 has a base sequence shown in the sequence ID. No. 17 adjacent to the 3' end of the exon 8, and has a base sequence shown in the sequence ID. No. 18 adjacent to the 5' end of the exon 9;

the intron intervening between exon 9 and exon 10 has a base sequence shown in the sequence ID. No. 19 adjacent to the 3' end of the exon 9, and has a base sequence shown in the sequence ID. No. 20 adjacent to the 5' end of the exon 10;

the intron intervening between exon 10 and exon 11 has a base sequence shown in the sequence ID. No. 21 adjacent to the 3' end of the exon 10, and has a base sequence shown in the sequence ID. No. 22 adjacent to the 5' end of the exon 11; and

the intron intervening between exon 11 and exon 12 has a base sequence shown in the sequence ID. No. 23 adjacent to the 3' end of the exon 11, and has a base sequence shown in the sequence ID. No. 24 adjacent to the 5' end of the exon 12.

12. A protein comprising 1 to 465 amino acid sequence in the sequence ID No. 1, or 1 to 437 amino acid sequence in the sequence ID. No. 2, and the protein being associated with Parkinson's disease, or having a substantially equivalent function thereto.

13. A protein expressed by the gene of any one of claims 1 to 4, the protein being associated with Parkinson's disease, or having an identical function thereto or a substantially equivalent function thereto.

14. A protein comprising an amino acid sequence translated by the gene of claim 5, and the protein being associated with Parkinson's disease, or having an identical function thereto or a substantially equivalent function thereto.

15. A protein comprising the amino acid sequence of claim 14 in which an amino acid is substituted, deleted, or added at least at one position, and the protein being associated with Parkinson's disease.

16. A protein (a) or (b):

(a) the protein comprising 1 to 465 amino acid sequence in the sequence ID. No. 1;

(b) the protein in which one or more amino acid(s) of the amino acid sequence is or are deleted, substituted, or added, and the protein being associated with Parkinson's disease.

17. A protein (c) or (d):

(c) the protein comprising 1 to 437 amino acid sequence

in the sequence ID. No. 2;

(d) the protein in which one or more amino acid(s) of the amino acid sequence is or are deleted, substituted, or added, and the protein being associated with Parkinson's disease.

18. A protein comprising the amino acid sequence of any one of claims 10 to 17 comprising:

a ubiquitin-like 1 to 72 amino acid sequence partially included in the sequence ID. No. 1; and

a zinc-finger-protein-like 418 to 449 amino acid sequence partially included in the sequence ID. No. 1.

19. A polypeptide or a protein consisting of a partial fragment of the amino acid sequence of any one of claims 10 to 17, or comprising the partial fragment thereof, or the full-length amino acid sequence thereof.

20. A monoclonal antibody or a polyclonal antibody against the protein of any one of claims 12 to 17.

21. A primer, or a probe, or an immobilized nucleic acid, or a DNA chip for use in detecting a base sequence, a genetic mutation, a deletion, and/or an expression amount of the DNA or the gene of any one of claims 1 to 8, or for use in

concentration thereof.

22. A primer, or a probe, or an immobilized nucleic acid, or a DNA chip for use in detecting a base sequence, a genetic mutation, a deletion, and/or an expression amount of RNA which is subjected to transcription and subjected to processing from the DNA or the gene of any one of claims 1 to 8, or for use in concentration thereof.

23. A primer, or a probe, or an immobilized nucleic acid, or a DNA chip for use in detecting a base sequence, a genetic mutation, and/or a deletion of the exon in the sequence ID No. 1 or No. 2, or for use in haplotyping a locus thereof.

24. A primer, or a probe, or an immobilized nucleic acid, or a DNA chip for use in detecting a base sequence, a genetic mutation, and/or a deletion of the intron of claim 11, or for use in haplotyping a locus thereof.

25. A primer or a probe for use in detecting a base sequence of the intron adjacent to the exon 1 of the gene being associated with Parkinson's disease of claim 1, or a locus thereof, the primer or probe comprising the following base sequence:

a base sequence of the sequence ID. No. 25 in the 5'-3'

direction of the sequence ID. No. 1 on the genome, and

a base sequence of the sequence ID. No. 26 in the 5'-3' direction on a complementary strand of the sequence ID. No. 1 on the genome.

26. A primer or a probe for use in detecting a base sequence of an intron adjacent to the exon 2 of the gene being associated with Parkinson's disease of claim 1, or a locus thereof, the primer or the probe comprising the following base sequence:

a base sequence of the sequence ID. No. 27 in the 5'-3' direction of the sequence ID. No. 1 on the genome, and

a base sequence of the sequence ID. No. 28 in the 5'-3' direction on a complementary strand of the sequence ID. No. 1 on the genome.

27. A primer or a probe for use in detecting a base sequence of an intron adjacent to the exon 3 of the gene being associated with Parkinson's disease of claim 1, or a locus thereof, the primer or the probe comprising the following base sequence:

a base sequence of the sequence ID. No. 29 in the 5'-3' direction of the sequence ID. No. 1 on the genome, and

a base sequence of the sequence ID. No. 30 in the 5'-3' direction on a complementary strand of the sequence ID. No. 1 on

the genome.

28. A primer or a probe for use in detecting a base sequence of the intron adjacent to the exon 4 of the gene being associated with Parkinson's disease of claim 1, or a locus thereof, the primer or probe comprising the following base sequence:

a base sequence of the sequence ID. No. 31 in the 5'-3' direction of the sequence ID. No. 1 on the genome, and

a base sequence of the sequence ID. No. 32 in the 5'-3' direction on a complementary strand of the sequence ID. No. 1 on the genome.

29. A primer or a probe for use in detecting a base sequence of an intron adjacent to the exon 4 of the gene being associated with Parkinson's disease of claim 1, or a locus thereof, the primer or the probe comprising the following base sequence:

a base sequence of the sequence ID. No. 33 in the 5'-3' direction of the sequence ID. No. 1 on the genome, and

a base sequence of the sequence ID. No. 34 in the 5'-3' direction on a complementary strand of the sequence ID. No. 1 on the genome.

30. A primer or a probe for use in detecting a base

sequence of an intron adjacent to the exon 5 of the gene being associated with Parkinson's disease of claim 1, or a locus thereof, the primer or the probe comprising the following base sequence:

a base sequence of the sequence ID. No. 35 in the 5'-3' direction of the sequence ID. No. 1 on the genome, and

a base sequence of the sequence ID. No. 36 in the 5'-3' direction on a complementary strand of the sequence ID. No. 1 on the genome.

31. A primer or a probe for use in detecting a base sequence of the intron adjacent to the exon 6 of the gene being associated with Parkinson's disease of claim 1, or a locus thereof, the primer or probe comprising the following base sequence:

a base sequence of the sequence ID. No. 37 in the 5'-3' direction of the sequence ID. No. 1 on the genome, and

a base sequence of the sequence ID. No. 38 in the 5'-3' direction on a complementary strand of the sequence ID. No. 1 on the genome.

32. A primer or a probe for use in detecting a base sequence of an intron adjacent to the exon 7 of the gene being associated with Parkinson's disease of claim 1, or a locus thereof, the primer or the probe comprising the following base

sequence:

a base sequence of the sequence ID. No. 39 in the 5'-3' direction of the sequence ID. No. 1 on the genome, and

a base sequence of the sequence ID. No. 40 in the 5'-3' direction on a complementary strand of the sequence ID. No. 1 on the genome.

33. A primer or a probe for use in detecting a base sequence of an intron adjacent to the exon 7 of the gene being associated with Parkinson's disease of claim 1, or a locus thereof, the primer or the probe comprising the following base sequence:

a base sequence of the sequence ID. No. 41 in the 5'-3' direction of the sequence ID. No. 1 on the genome, and

a base sequence of the sequence ID. No. 42 in the 5'-3' direction on a complementary strand of the sequence ID. No. 1 on the genome.

34. A primer or a probe for use in detecting a base sequence of the intron adjacent to the exon 8 of the gene being associated with Parkinson's disease of claim 1, or a locus thereof, the primer or probe comprising the following base sequence:

a base sequence of the sequence ID. No. 43 in the 5'-3' direction of the sequence ID. No. 1 on the genome, and

a base sequence of the sequence ID. No. 44 in the 5'-3' direction on a complementary strand of the sequence ID. No. 1 on the genome.

35. A primer or a probe for use in detecting a base sequence of an intron adjacent to the exon 9 of the gene being associated with Parkinson's disease of claim 1, or a locus thereof, the primer or the probe comprising the following base sequence:

a base sequence of the sequence ID. No. 45 in the 5'-3' direction of the sequence ID. No. 1 on the genome, and

a base sequence of the sequence ID. No. 46 in the 5'-3' direction on a complementary strand of the sequence ID. No. 1 on the genome.

36. A primer or a probe for use in detecting a base sequence of an intron adjacent to the exon 10 of the gene being associated with Parkinson's disease of claim 1, or a locus thereof, the primer or the probe comprising the following base sequence:

a base sequence of the sequence ID. No. 47 in the 5'-3' direction of the sequence ID. No. 1 on the genome, and

a base sequence of the sequence ID. No. 48 in the 5'-3' direction on a complementary strand of the sequence ID. No. 1 on the genome.

37. A primer or a probe for use in detecting a base sequence of the intron adjacent to the exon 11 of the gene being associated with Parkinson's disease of claim 1, or a locus thereof, the primer or probe comprising the following base sequence:

a base sequence of the sequence ID. No. 49 in the 5'-3' direction of the sequence ID. No. 1 on the genome, and

a base sequence of the sequence ID. No. 50 in the 5'-3' direction on a complementary strand of the sequence ID. No. 1 on the genome.

38. A primer or a probe for use in detecting a base sequence of an intron adjacent to the exon 12 of the gene being associated with Parkinson's disease of claim 1, or a locus thereof, the primer or the probe comprising the following base sequence:

a base sequence of the sequence ID. No. 51 in the 5'-3' direction of the sequence ID. No. 1 on the genome, and

a base sequence of the sequence ID. No. 52 in the 5'-3' direction on a complementary strand of the sequence ID. No. 1 on the genome.

39. A primer or a probe of any one of claims 24 to 38 for use in detecting a base sequence of the intron adjacent to

the exon of the gene being associated with Parkinson's disease, or a locus thereof.

40. An oligonucleotide, or an oligonucleotide analog, or a modified product thereof which comprises a partial sequence of the base sequence of any one of claims 1 to 8, or which is hybridizable with the base sequence of any one of claims 1 to 8.

41. An oligonucleotide for use in amplifying the full-length base sequence or the partial base sequence of any one of claims 1 to 8, or the oligonucleotide for use in amplifying partially the full-length base sequence or the partial base sequence of any one of claims 1 to 8, according to PCR method using a human RNA as a template, PCR method or RT-PCR method using a human cDNA as a template.

42. An oligonucleotide for use in amplifying the base sequence comprising the exon in the sequence ID. No. 1 or No. 2 and the intron of claim 11 which is adjacent to the exon according to PCR method, or the oligonucleotide for use in amplifying a part of the base sequence according to PCR method.